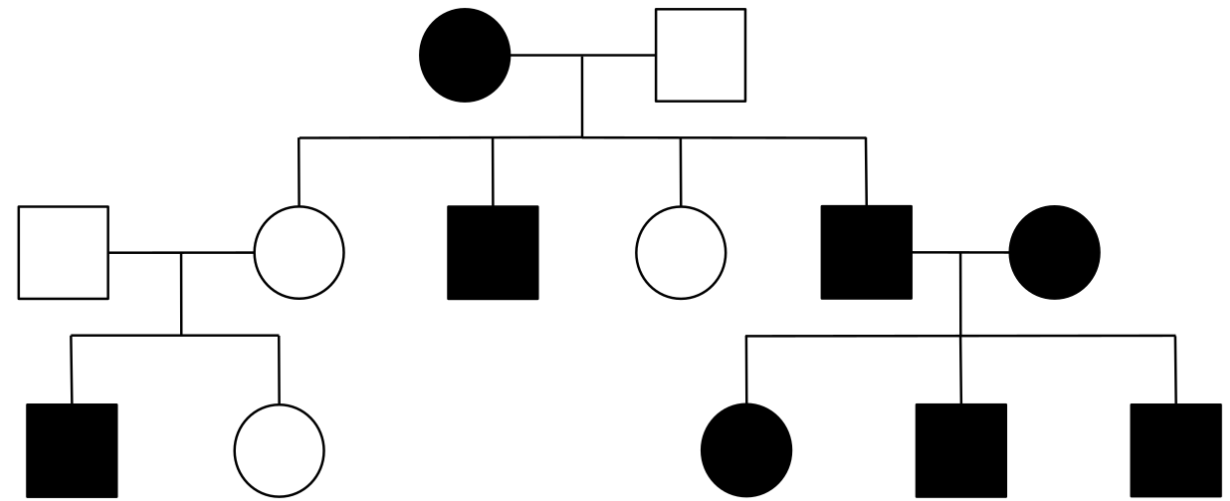


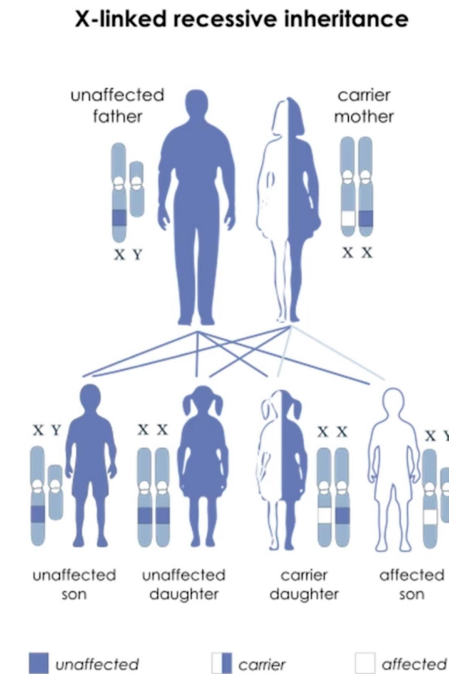
Lesson 16

Pedigrees



Pedigrees

- Sometimes, the genetics of a particular trait are very complicated
 - There might be many genes involved
 - There might be traits that are associated with sex
 - There might be traits that are influenced in ways that we just don't understand
- **Pedigrees** = another set of tools to understand complex traits
 - Especially important to understand diseases



Pedigree analysis
uncovers type of inheritance
suggests gene(s) characteristics

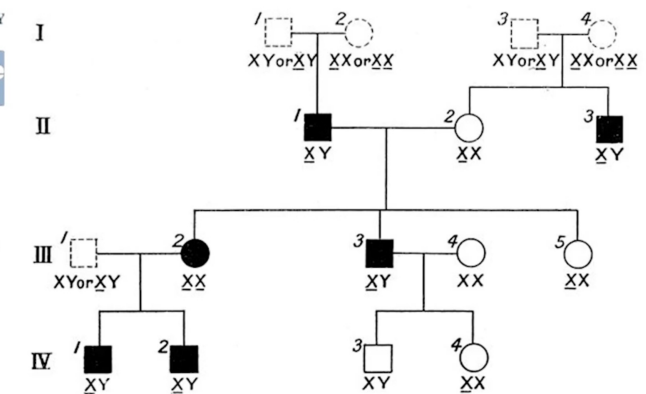


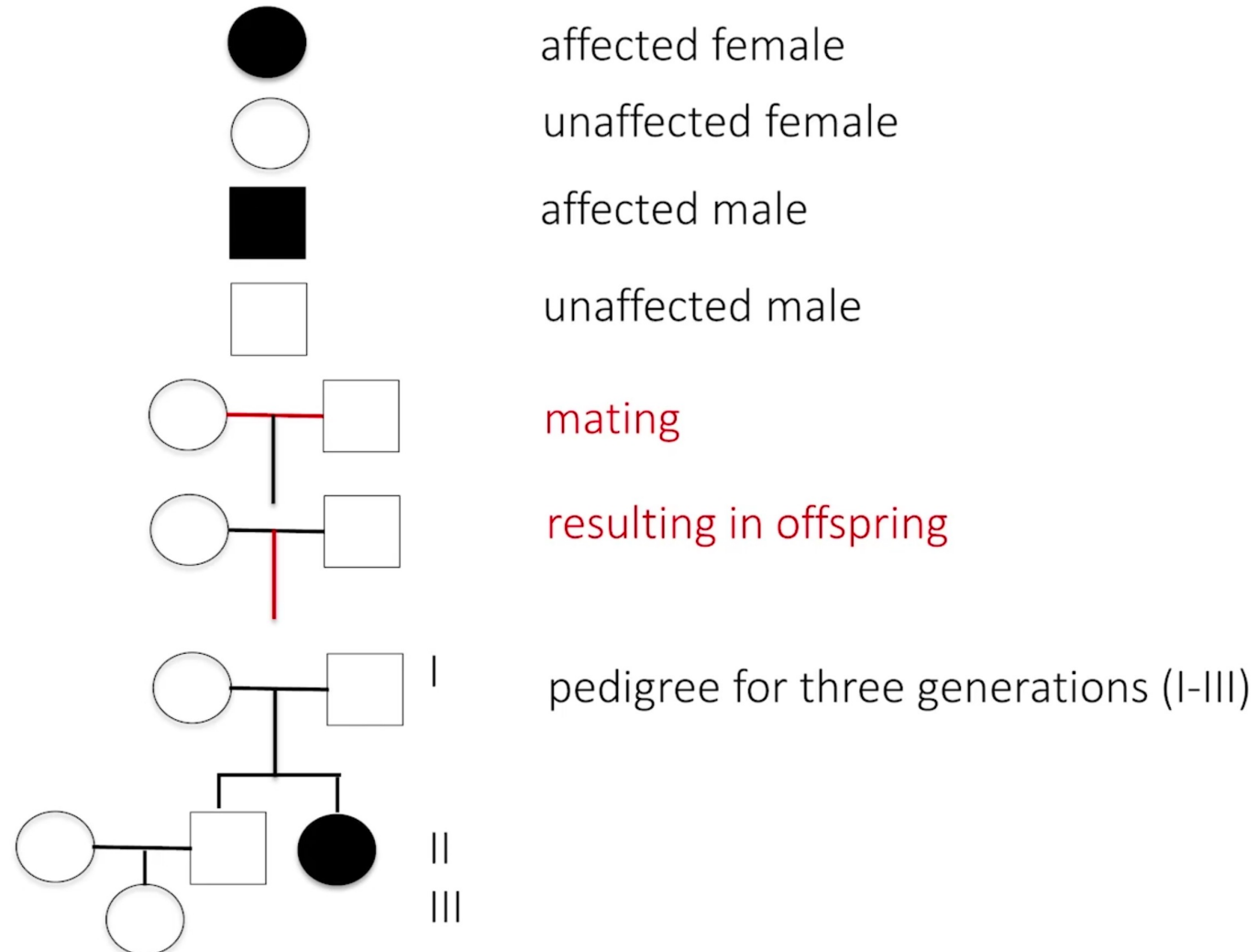
FIG. 21.—PEDIGREE CHART OF COLOUR-BLINDNESS.

Pedigrees

- **Pedigrees = genetics from family history**
- Conventions in pedigree writing:
 - ○ female
 - □ male
 - ● ■ the trait you are looking at is present (*e.g.*, a disease, brown eyes, etc.)

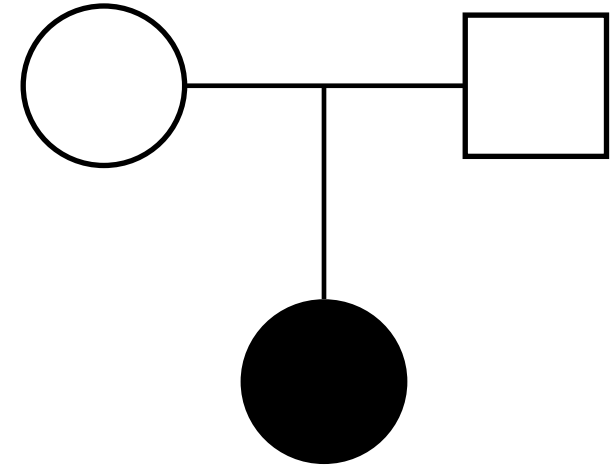
More pedigree nomenclature

Pedigree nomenclature



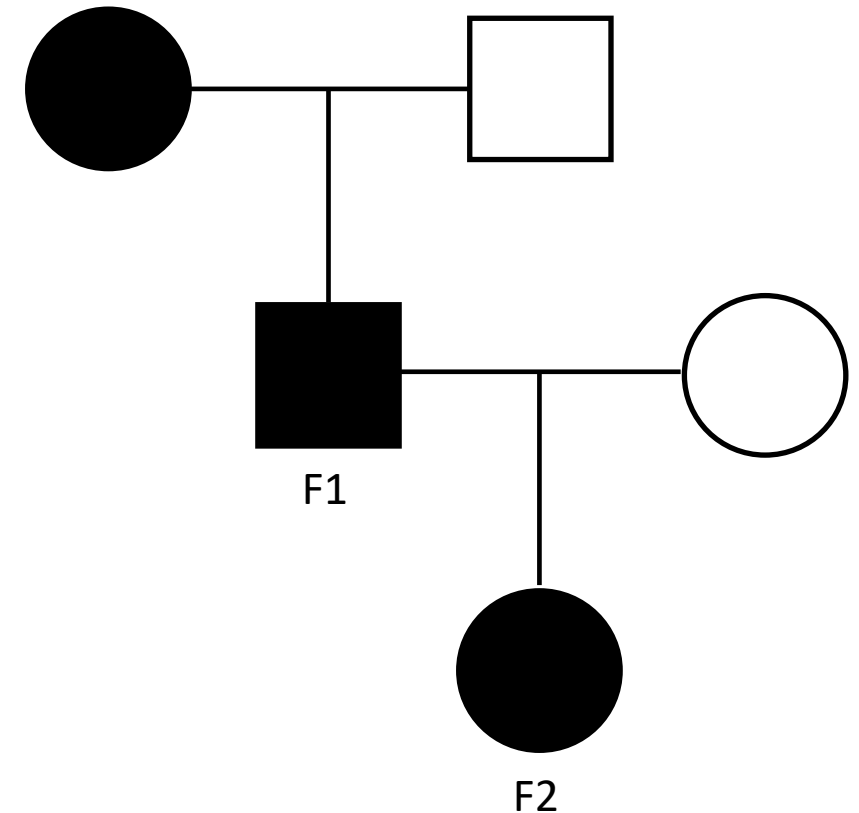
Pedigrees- 1

- This pedigree (*e.g.*, disease):
 - Two healthy parents
 - One affected daughter
- **This is a characteristic of a recessive trait**



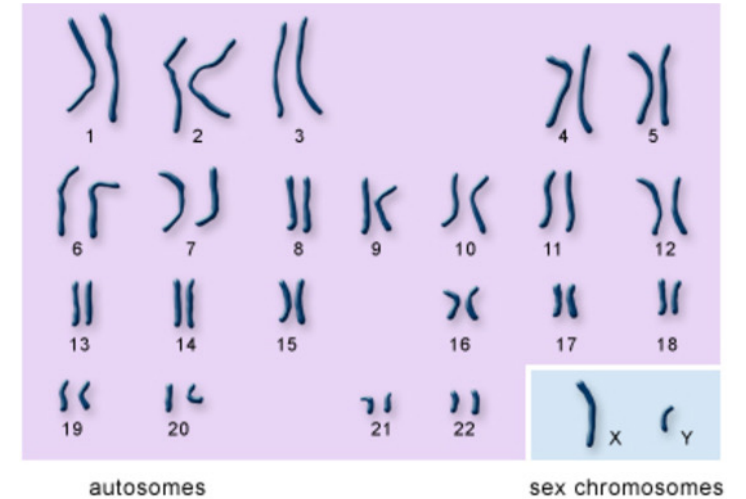
Pedigrees - 2

- This pedigree (*e.g.*, disease):
 - One affected parent and one unaffected parent (*e.g.*, father)
 - One affected son (F1)
 - The affected son mates an unaffected female
 - One affected daughter (F2)
- **The pedigree pattern where every affected offspring has an affected parent is characteristic of a dominant trait**



Chromosomes and pedigrees

- Eukaryote cells have two types of chrs:
 - Autosomes = 22 paired chrs (2 chrs 1, 2 chrs2, etc....)
 - Sex chrs*:
 - XX (paired) → female * simplified
 - XY (unpaired) → male



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- 3 types of pedigree based on autosomes or sex chromosomes:
- Autosomal recessive } Males and females affected equally
- Autosomal dominant }

Chromosomes and pedigrees

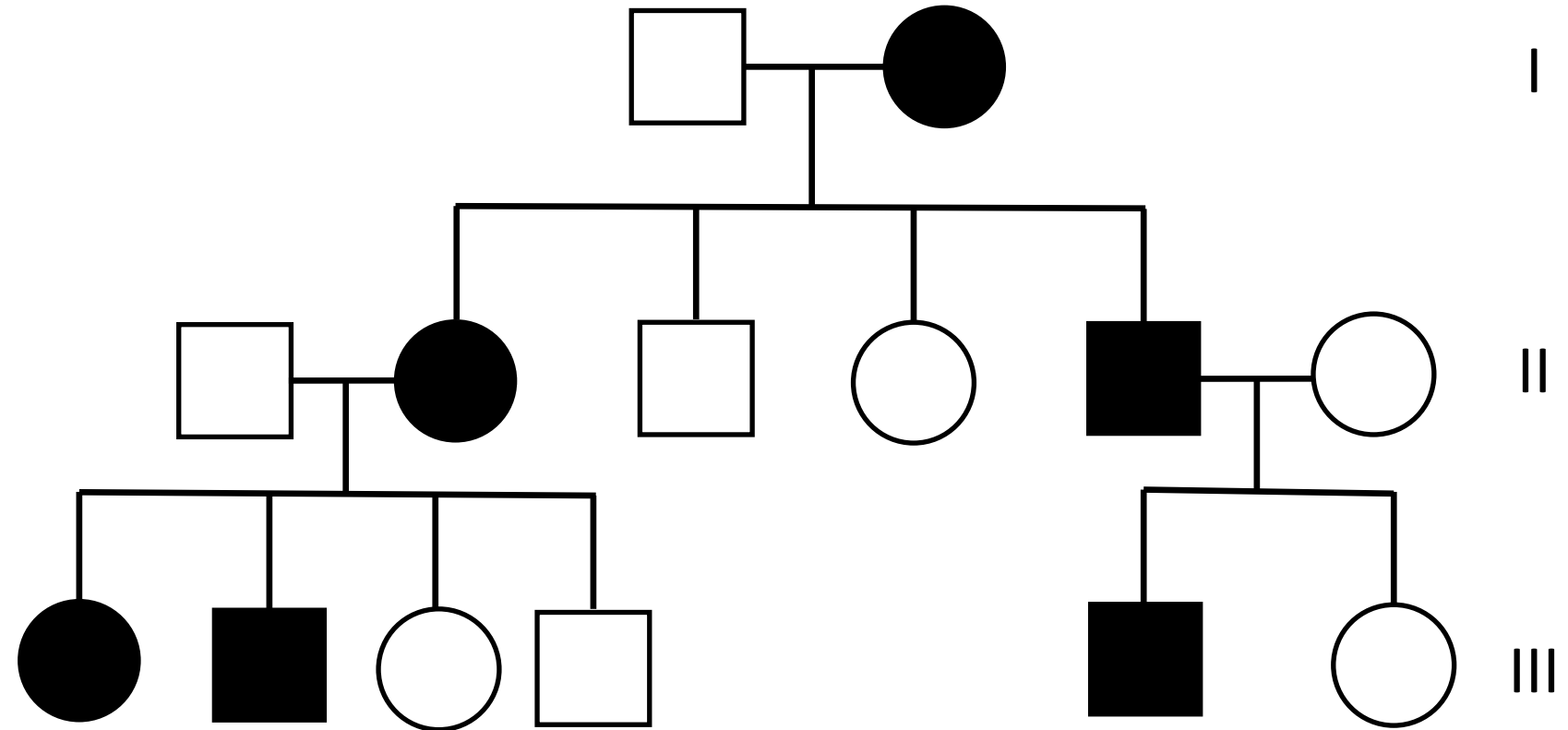
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- 3 types of of pedigree based on autosomes or sex chromosomes:
 - Autosomal recessive
 - Autosomal dominant } Males and females affected equally
- **X-linked recessive → Males affected more than females**
 - there is only one X (the other allele is Y) and is affected → the trait will show itself)
 - It is X-linked recessive because this trait on the X allele is always transmitted to the SONS (XY) from the mother

Pedigree for autosomal dominant trait

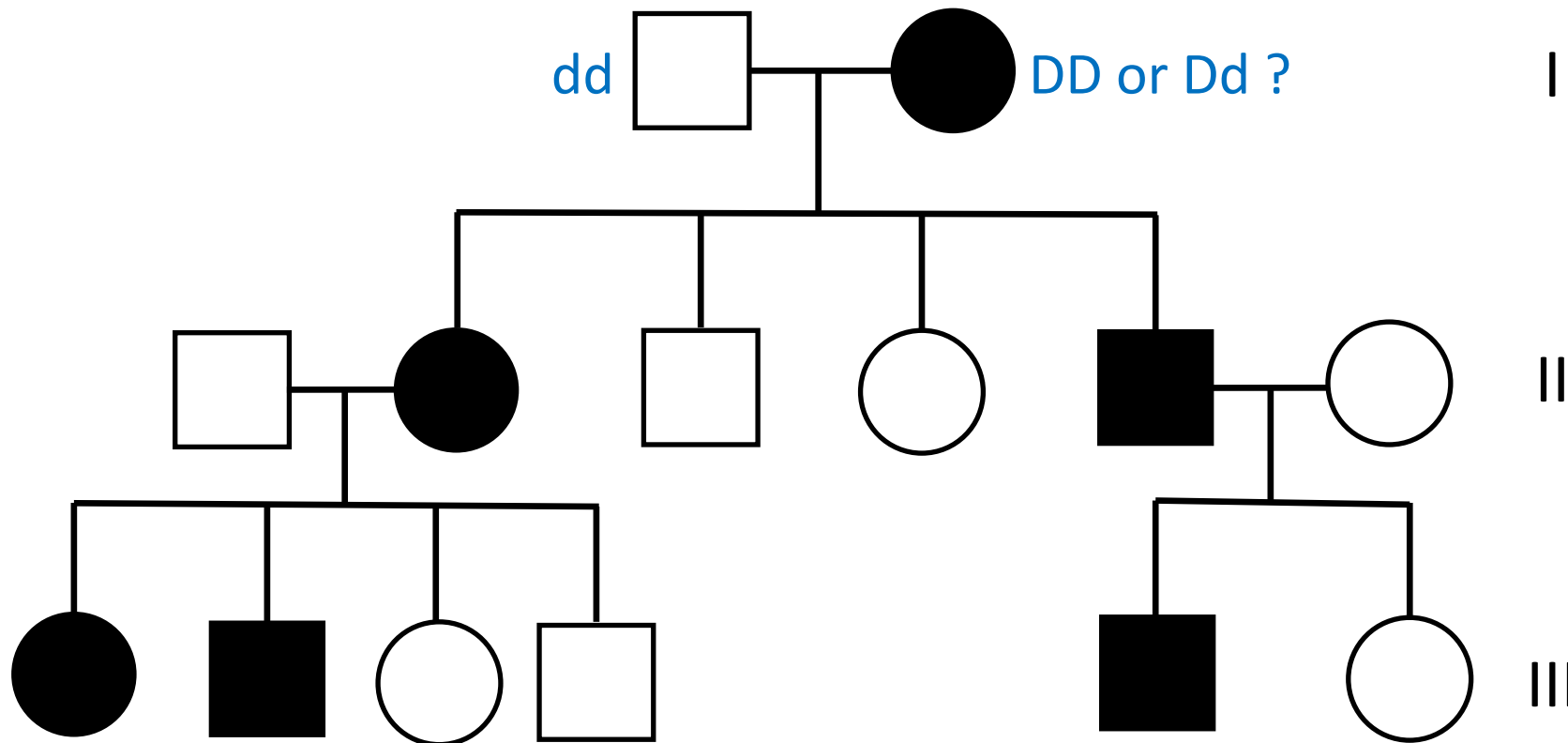
Clues

Autosomal: both males and females are affected



Dominant: every affected child has one affected parent



Pedigree for autosomal dominant trait (with genotypes)

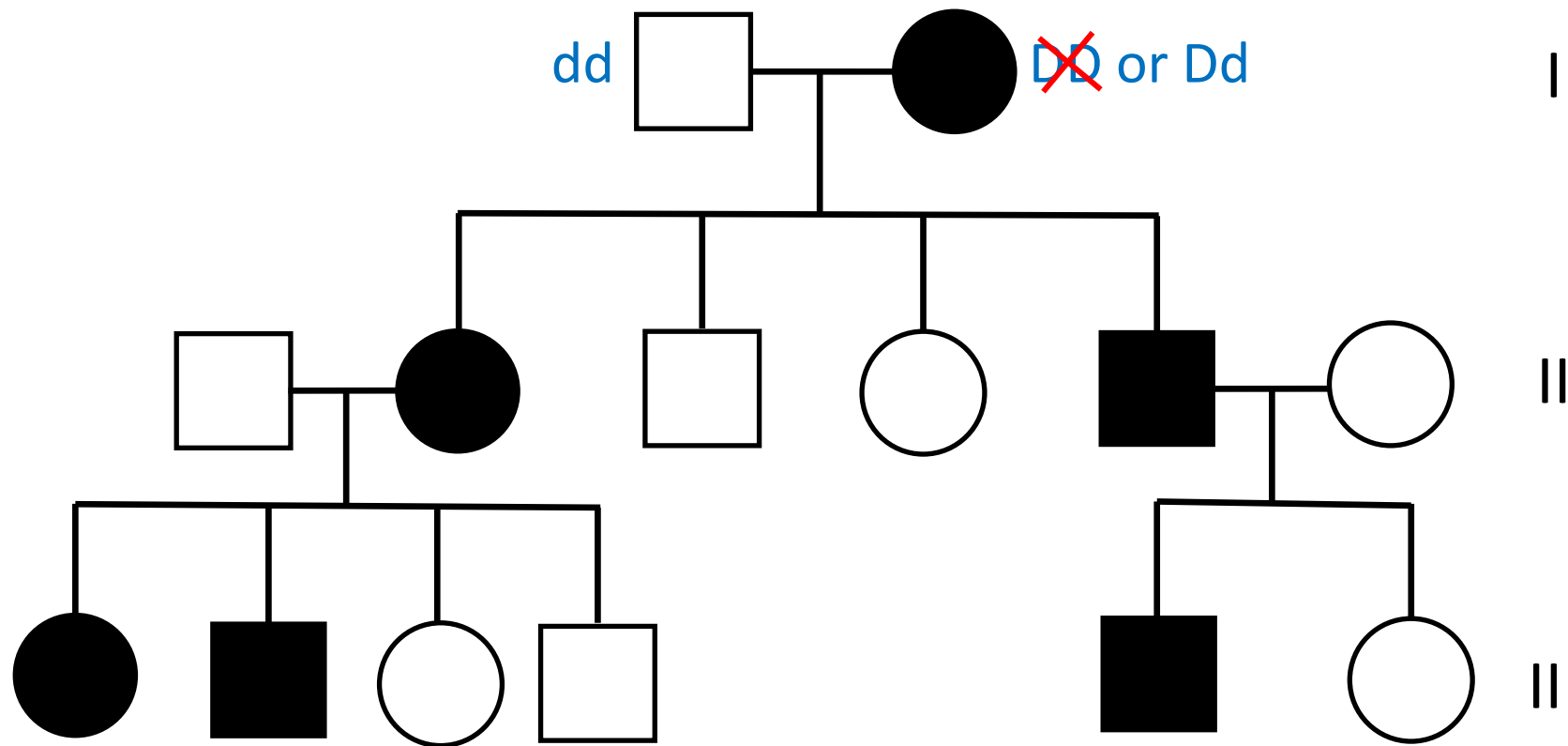


Punnett squares for gen II (gen 1 = parents)



		Gametes	
		D	D
G a m e t e s	d	Dd	Dd
	d	Dd	Dd

100% of II would be affected (all have the dominant allele D)



Pedigree for autosomal dominant trait (with genotypes)



Punnett squares for gen II (gen I = parents)

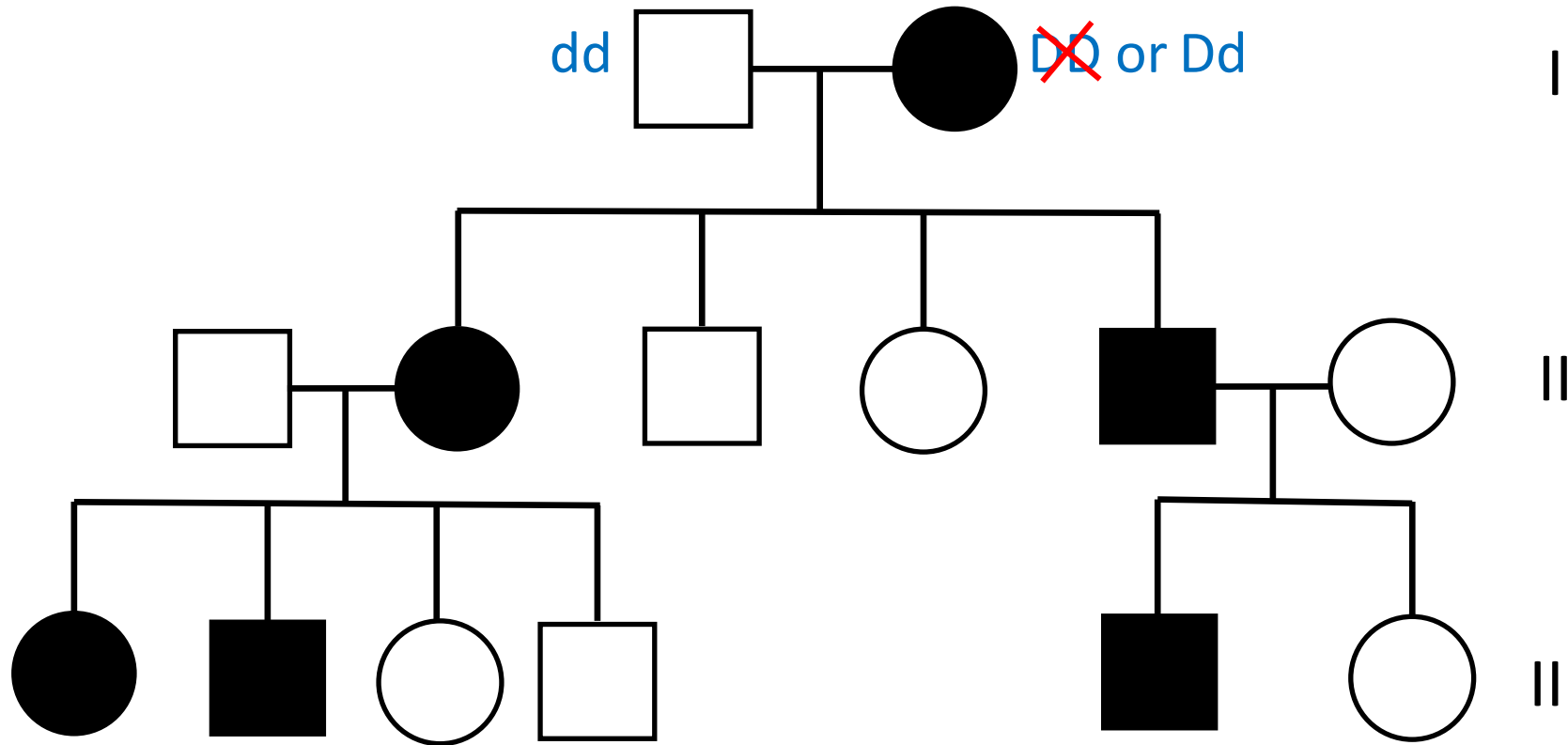
		Gametes	
		D	D
G a m e t e s	d	Dd	Dd
	d	Dd	Dd

100% of F1 would be affected (all have the dominant allele D)

		Gametes	
		D	d
G a m e t e s	d	Dd	dd
	d	Dd	dd

50% of II would be affected (half have the dominant allele D)

Pedigree for autosomal dominant trait (with genotypes)



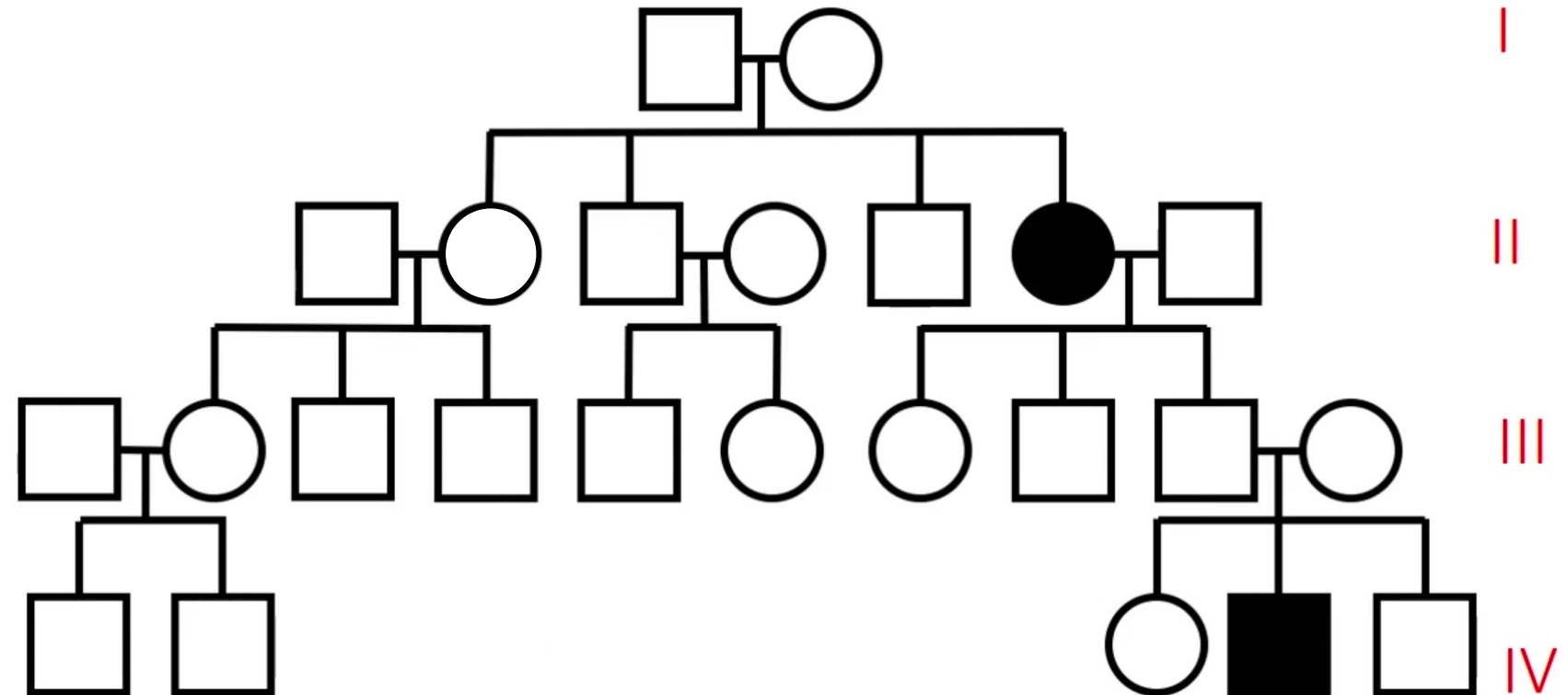
Note: you may not always get ratios identical to predicted*

Pedigree for autosomal recessive trait

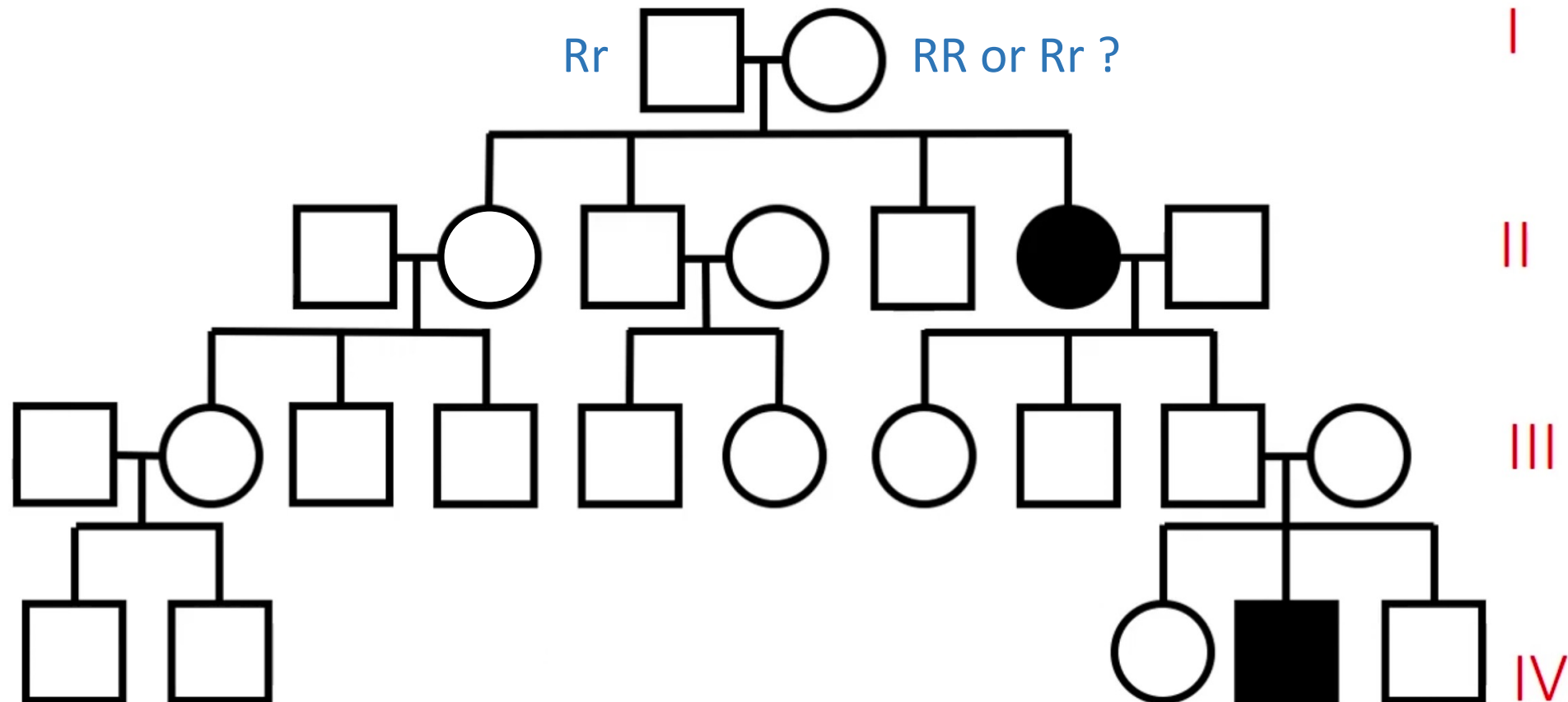
Clues

Autosomal: both males and females are affected

Recessive: affected child(ren) from unaffected parents
not many affected offspring

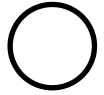



Pedigree for autosomal recessive trait (with genotypes)



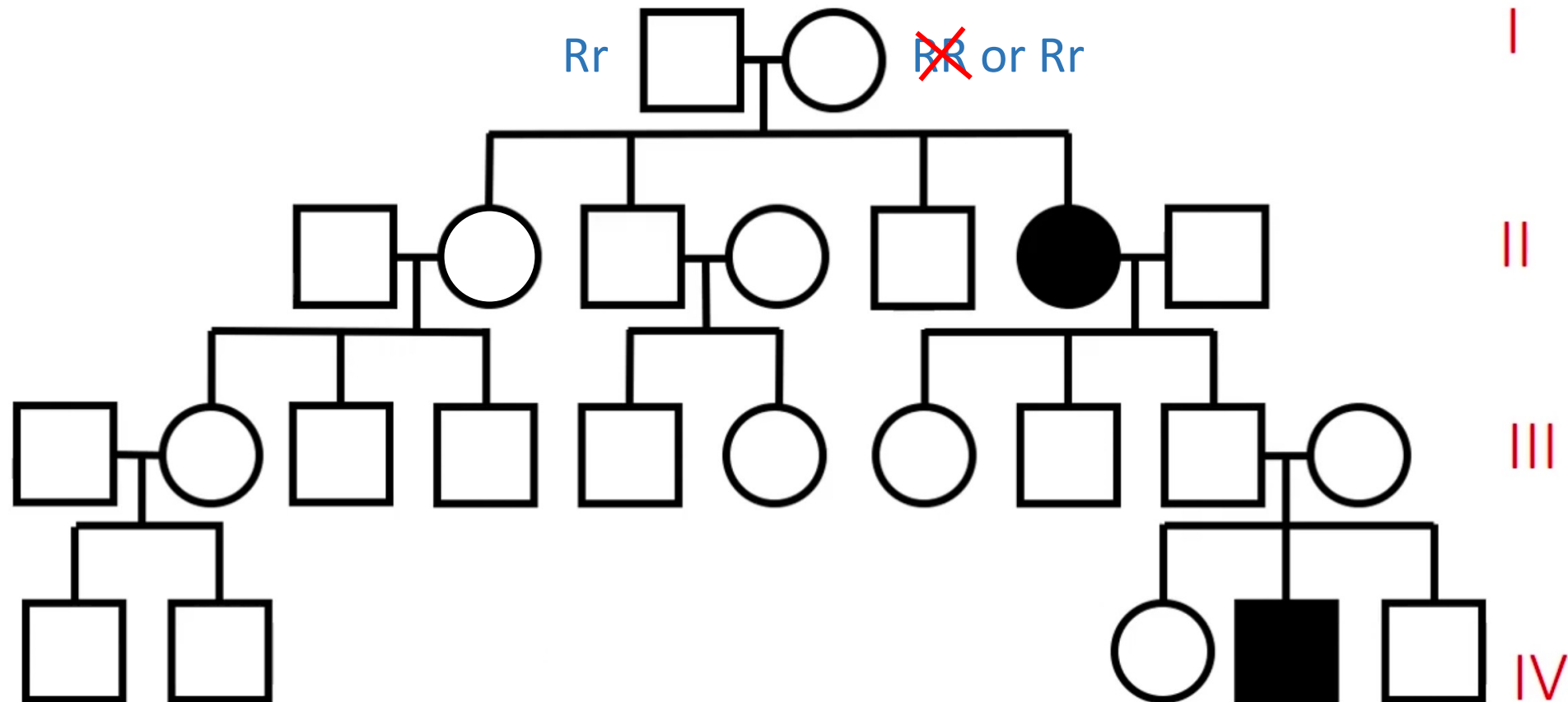
Remember: it is the recessive allele r that expresses the disease

Punnett squares for gen II (gen 1 = parents)



		Gametes	
		R	R
G a m e t e s	R	RR	RR
	r	Rr	Rr

0% of II would be affected (none has genotype rr)



Pedigree for autosomal recessive trait (with genotypes)



Punnett squares for gen II (gen 1 = parents)

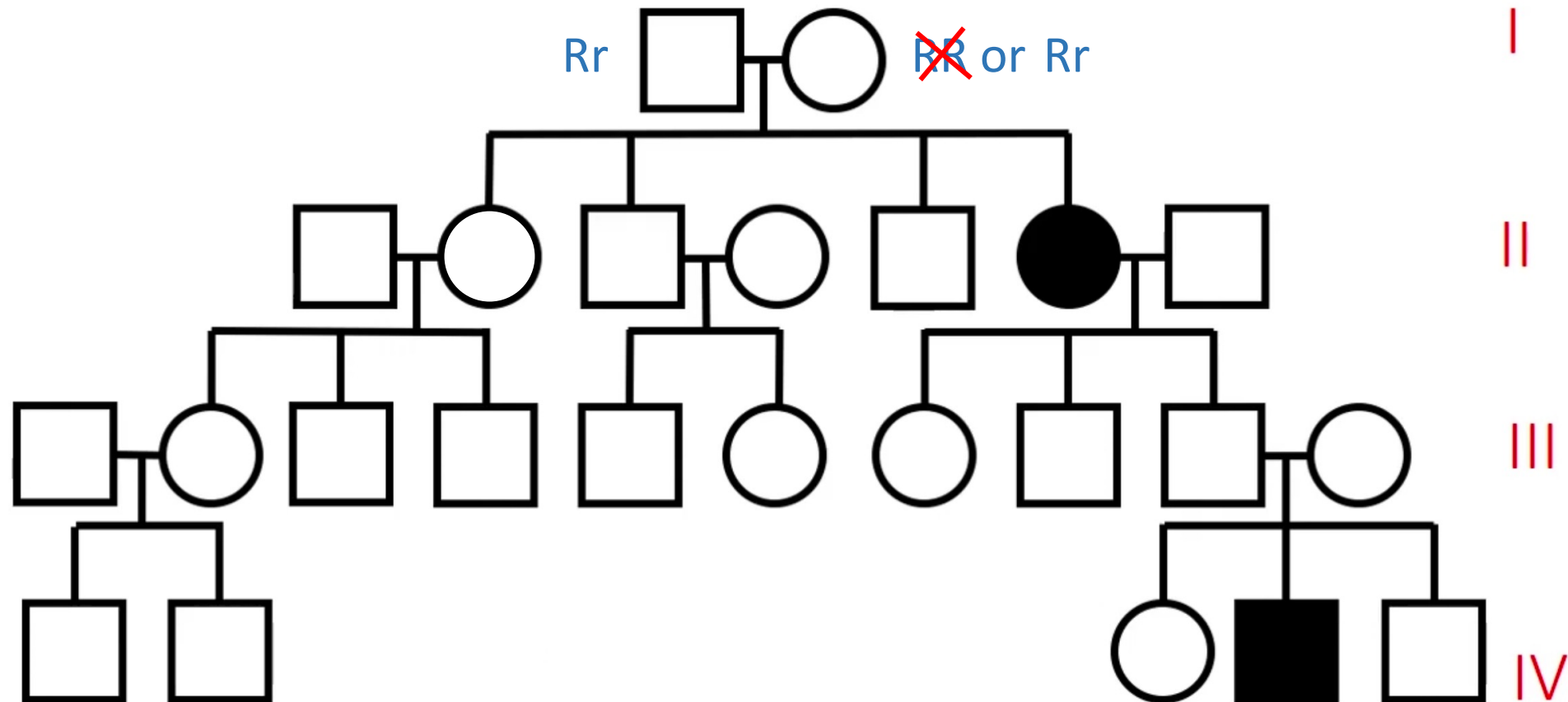
			Gametes	
		R	R	
G a m e t e s	R	RR	RR	
	r	Rr	Rr	

0% of II would be affected (none has genotype rr)

			Gametes	
		R	r	
G a m e t e s	R	RR	Rr	
	r	Rr	rr	

25% of II (1:3) would be affected (only 1 has rr)

Pedigree for autosomal recessive trait (with genotypes)



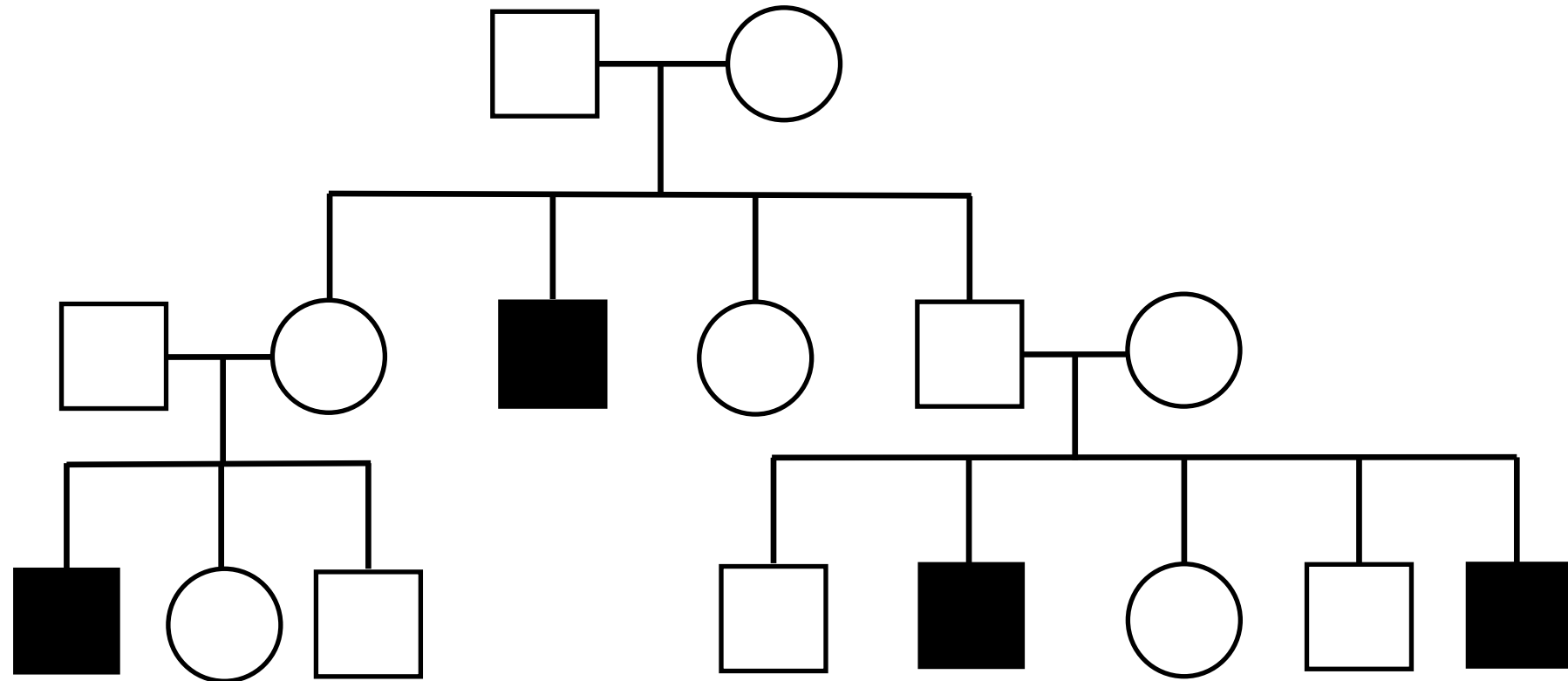
Note: you may not always get ratios identical to predicted*

Pedigree for X-linked recessive trait

Clues

X-linked: only males affected

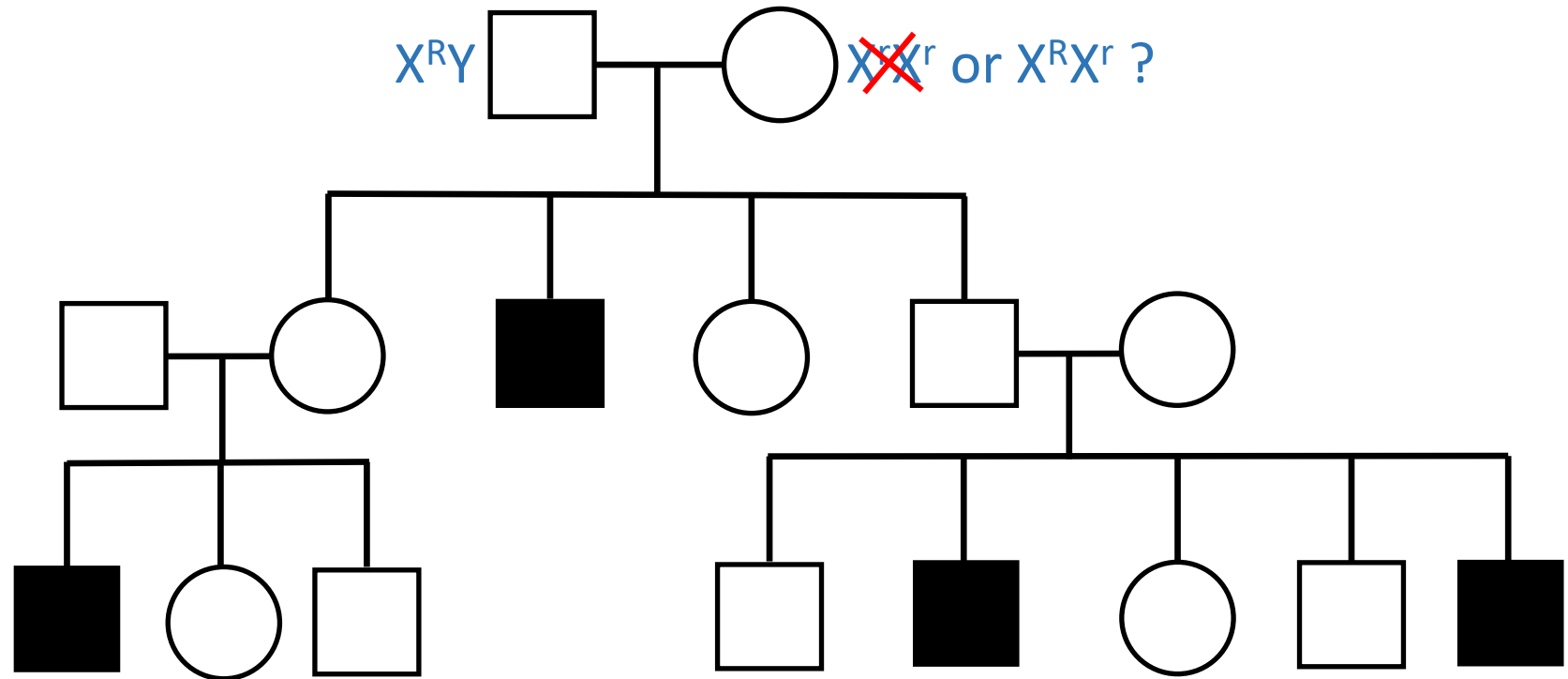
Recessive: affected child(ren) from unaffected parents
not many affected offspring





Pedigree for X-linked recessive trait (with genotypes)

Remember: it is the recessive allele r that expresses the disease

X^rX^r can be immediately excluded otherwise the mother would have the trait

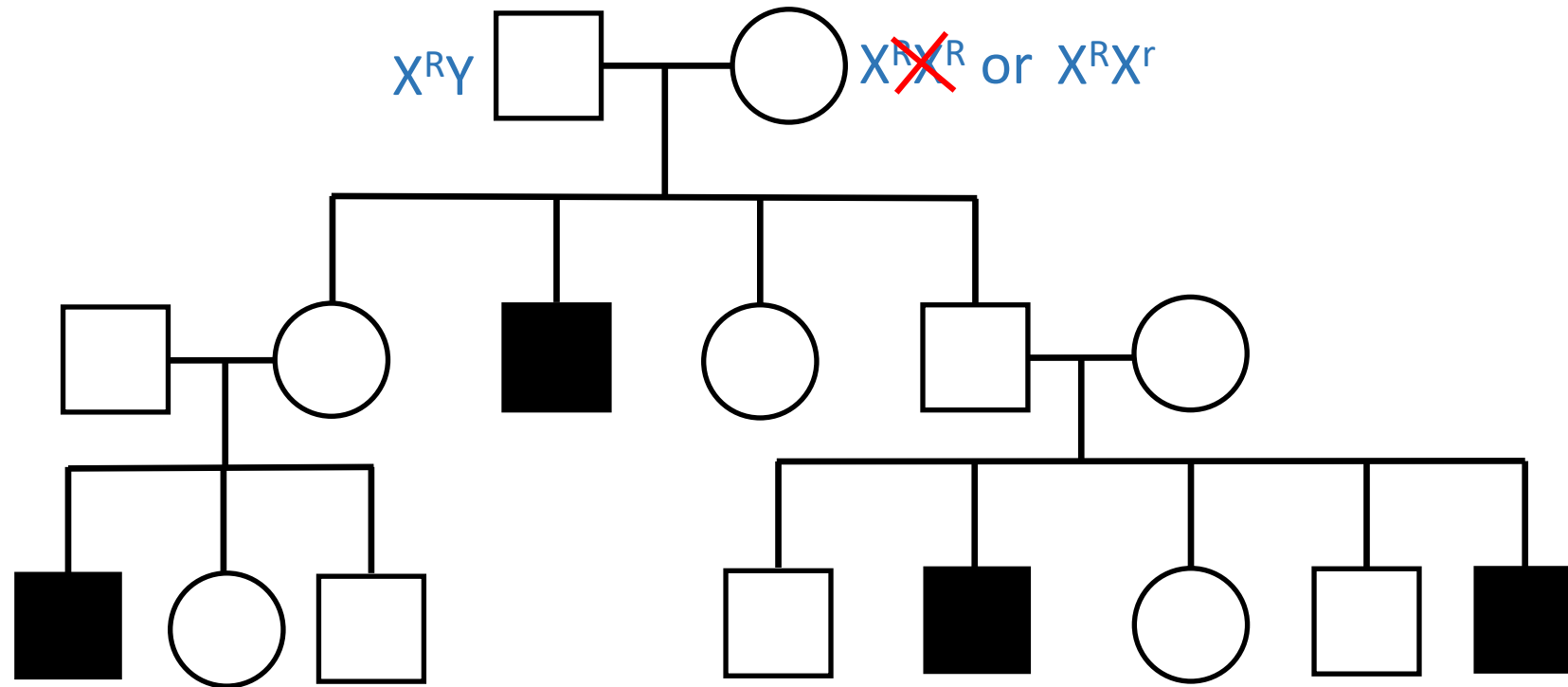


Punnett squares for gen II (gen 1 = parents)

		Gametes	
		X^R	X^r
G a m e t e s	X^R	$X^R X^R$	$X^R X^r$
	Y	$X^R Y$	$X^r Y$

50% of MALE II (1:1) would be affected (has genotype $X^r Y$)

Pedigree for X-linked recessive trait (with genotypes)



Note: you may not always get ratios identical to predicted*

Notes

- The discussed pedigrees are simple and ideal
- You may not always get ratios identical to predicted due to non-Mendelian inheritance patterns, which include (among others):
 - **Codominance and incomplete dominance**
 - **Polygenic traits**: some traits are determined by the combined effect of more than one pair of genes → polygenic (or continuous) traits (an example of this is human stature)
 - **Multiple-allele series**: the ABO blood type system is also an example of a trait that is controlled by more than just a single pair of alleles
 - **Modifying and regulator genes**: modifying genes alter how certain other genes are expressed in the phenotype (for instance, here is a dominant cataract gene which will produce varying degrees of vision impairment depending on the presence of a specific allele for a companion modifying gene)

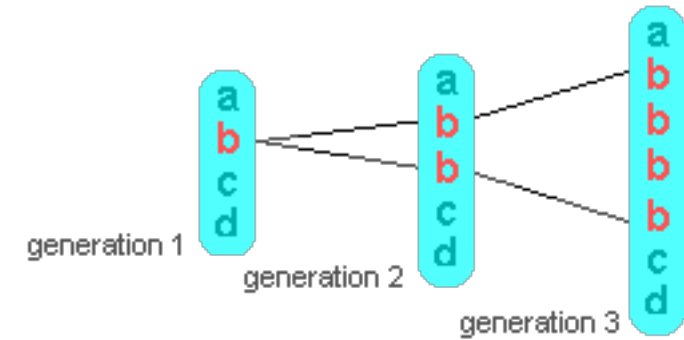
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 - their effect does not normally occur unless certain environmental factors are present. For example:
 - you may inherit the genes that are responsible for type 2 diabetes but never get the disease unless you become greatly overweight, persistently stressed psychologically, or do not get enough sleep on a regular basis;
 - the genes that cause the chronic autoimmune disease, multiple sclerosis may be triggered by the Epstein-Barr virus and possibly other specific environmental stresses

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- **Pleiotropy**: a single gene may be responsible for a variety of traits. The complex of symptoms that are collectively referred to as sickle-cell trait or sickle-cell anemia, is an example.
 - A single gene results in irregularly shaped red blood cells that painfully block blood vessels, cause poor overall physical development, as well as related heart, lung, kidney, and eye problems

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 - **Stuttering alleles:** some genetically inherited diseases have more severe symptoms each succeeding generation due to segments of the defective genes being doubled in their transmission to children (typically fatal degenerative nerve disorders)
- You may not have enough offspring at F1 to interpret pedigree
 - You need to analyze F2, F3, ...